29th Annual Meeting of
The European Society of Human Genetics

Board of the ESHG 1996/97

S. Aymé, President (Villejuif) • *A. Schinzel,* Vice-President (Zurich) • *M. A. Ferguson-Smith,* President Elect (Cambridge) • *J.J. Cassiman,* Secretary General (Leuven) • *A. Ballabio* (Milan) • *J. Burn* (Newcastle upon Tyne) • *C. Delozier-Blanchet* (Geneva) • *M. Devoto* (Genoa) • *X. Estivill* (Barcelona) • *H. Galjaard* (Rotterdam) • *P.A. Jacobs* (Salisbury) • *S. Lyonnet* (Paris) • *K. Méhes* (Pecs) • *K. Orstavik* (Oslo) • *L. Peltonen* (Helsinki) • *M.E. Pembrey* (London) • *M.B. Petersen* (Athens) • *M. Schwartz* (Copenhagen) • *K. Sperling* (Berlin) • *N. Tommerup* (Glostrup) • *C. Van Broeckhoven* (Antwerp) • *G.J.B. van Ommen* (Leiden)

Scientific Programme Committee of the ESHG

C.H.C.M. Buys, Chair (Groningen) • *M. Vekemans,*Vice Chair (Paris) • *S. Antonarakis* (Geneva) • *A. Ballabio* (Milan) • *M. Devoto* (Genoa) • *H. Santos* (Lisbon) • *P. Farndon* (Birmingham) • *T. Meitinger* (Munich) • *C. Monteiro* (Lisbon) • *A. Read* (Manchester) • *G. Romeo* (Genoa and Lyon) • *C. Van Broeckhoven* (Antwerp) • *R. M. Winter* (London)

Organizing Committee

F. Ajmar (Genoa), *A. Ballabio* (Milan), *G. Camera* (Genoa), *B. Dallapiccola* (Rome), *F. Dagna Bricarelli* (Genoa), *M. Devoto* (Genoa), *G. Romeo* (Genoa and Lyon), *L. Varesio* (Genoa)

Scientific Secretariat
Lab. of Molecular Genetics • Istituto G. Gaslini
16148 Genova - Italy
Phone: +39 10 5636370/400/532
Fax: +39 10 3779797
e-mail: genetica@unige.it

Meetings Organizer
Symposia Snc Via A. Cecchi 7/12
16129 Genova - Italy
Phone: +39 10 5704092/3
Fax: +39 10 5704093
e-mail: symposia@panet.it

In collaboration with
Direzione Scientifica Istituto G. Gaslini - Genova

The Web site of ESHG97 and this book of Abstracts were prepared by Ing. Luigi Marras
(gmarras@mbox.vol.it • gmarras@tin.it)
60% of the Abstracts were received in electronic format through the Web site

cancer sample. In particular, a sense of guilt about transmission of susceptibility to breast cancer to daughters it was often observed in this group; 2) it was important to adapt the comunication with patients to their own communicative style and subjective characteristics, cultural customs, values, intellective level and age. We suggest that written educational material should be provided in order to improve the understanding of basic concepts like: research, genetics, prevention, risk. In doing this, psychological and social variables must be take into account.

This work was supported by AIRC.

P7.005

Certification of master's level genetic counselors and accreditation of genetic counseling graduate programs: a model for quality assurance.

Fine, Beth (1), and Baker, D. (2)
(1) Northwestern University Medical School, Chicago IL, USA; (2) University of Michigan, Ann Arbor MI, USA

Medical genetics practice in the USA is a team effort, providing diagnosis, management and counselling to individuals and families within a client-centred ethnocultural context. Master's level genetic counsellors are trained in human/medical genetics, psychology, communication skills and ethics to bridge the gap between the laboratory and the patient. Until 1993, the American Board of Medical Genetics certified geneticists with doctoral or master's degrees. The American Board of Genetic Counselling (ABGC) was then formed to establish criteria and procedures for certification of master's level genetic counsellors and accreditation of graduate programs. Its goal is to improve the quality of education, reflecting changes in genetic technology. Certification promotes the public welfare by ensuring a minimum level of practitioner competence. The ABGC has taken an innovative, integrated approach to certification and accreditation by creating practice-based competencies to define and evaluate training outcome measures. We will describe the methodology for developing the competencies; a protocol for program accreditation, and, the credentialing and certification criteria, and evaluation process. The impact of the ABGC's efforts on professional advancement and on programs will be discussed. We will conclude with the ABGC's future goals and recommendations for adapting this credentialing model for other genetic health care systems.

P7.006

Organization of genetic counseling in Italy: input of psychosocial disciplines.

Jacopini, Gioia (1); Frontali, M.(2); Perobelli S.(3), and Salmaso D.(1)
(1) Istituto di Psicologia CNR, Roma, Italy; (2) Istituto di Medicina Sperimentale CNR, Roma, Italy; (3) Servizio Psicologia Clinica, C. Fibrosi Cistica Verona, Italy

Developments in genetics have expanded predictive and diagnostic potential with respect to an increasing number of diseases. This body of knowledge, however, places the individual before the need to make increasingly complex choices which deepen the psychosocial problems that genetic counselors have to be able to give answers to.

In Italy there are various genetic diagnostic and counseling centres even though they are not uniformly distributed throughout the Country. In order to gain an understanding of how many of these centres are truly capable of facing the complex psychosocial problems posed by genetic counseling and, in particular, by the management of diagnostic or predictive testing we have drawn up a questionnaire and sent it out to the public and private centres listed in the Guide to the Diagnostic and Treatment Services for Genetic Disorders set up by AIRH and Ministry for Health.

Preliminary data trace a picture of severe delay with regard to the care for the psychosocial components of genetic counseling.

If the final data will confirm this picture, it is most urgent that Health policy makers in Italy be made aware of the danger of the spread of genetic diagnoses throughout the country without there being adequate psychosocial support.

P7.007

The role of the non-medical genetic counsellor in cancer referrals to a Regional Genetic Service

MacLeod, Rhona; Clancy, T.; Durell, S.; Faulkner, C.; Hall, G., and Kerzin-Storrar, L.
St Mary's Hospital, Manchester, UK.

The increasing number of cancer referrals to genetics departments has led to an exciting challenge for non-medical genetic counsellors in the setting up of their own clinics, managing registers for cancer predisposing conditions, involvement in pre-symptomatic test counselling, and educating other health professionals. This paper will describe the work of genetic associates (non-medical genetic counsellors and nurses) in Manchester, UK.

A Genetic Register Service has been in existence for families with dominant tumour predisposing conditions, eg VHL, FAP and NF2, since 1990. The responsibilities of the genetic associate in managing the register include: coordinating screening for early detection of problems; offering genetic counselling to the extended family; and pre-symptomatic test counselling.

A genetic associate and a clinical geneticist are both involved in the counselling for pre-symptomatic testing. At least one session is with the genetic associate only. This multidisciplinary approach is modelled on our protocol for HD testing.

The education of other health professionals about cancer genetics is vital if the service is not to be overwhelmed by inappropriate referrals. The content of a workshop for practice nurses in the North West of England will be outlined.

P7.008

Defining the skills and knowledge base of genetic nursing practice

Penny, Guilbert
Department of Clinical Genetics, City Hospital, Nottingham, UK

The Delphi technique was used to determine the opinions of an expert practitioner panel regarding the core components of genetic practice and the knowledge base skills and personal attributes required to deliver an optimum service. The items generated by the panel were the subject of iterative review to establish a consensus view. 96 5 consensus was achieved over three rounds,. The main components of practice identified included interaction with clients involving information transfer and counselling around issues concerning psychosocial impact. Educational and research components of practice were also valued highly. Commensurate knowledge and skills were identified to support its practice most of which were considered to be embedded in prior clinical experience in other settings which could be developed as a practitioner in the speciality. Academic study to at least first degree level with the goal of a Masters degree was identified. This would fit in with the designated UKCC education requirements for specialist and advanced practitioners. The results of the study can be used as a basis upon which to develop appropriate specialist educational programmes for future practitioners and to define competencies and standards which could be used to guide specification and appraisal of nursing contributions to genetic services.

29TH Annual Meeting of the
EUROPEAN SOCIETY OF HUMAN GENETICS
May 18-20, 1997, Genoa, Italy

Satellite Meeting on:

Education, Training and Responsibilities of Non-Medical Genetic Counsellors
May 17, 1997, Magazzini del Cotone (Scirocco Hall), Genoa, Italy.

Program

08:30-09:00 Registration
09:00-09:30 Welcome address
09;30-13:00 Invited speakers
13:00-14:30 Lunch
14:30-15:30 Invited speakers
15:30-16:30 Communications
16:30-17:30 Round Table
18:00-20:00 Visit to the van Dyck special exibition (Palazzo Ducale-Genoa)
20:00 Welcome party of ESHG meeting

Speakers

Ségolène Aymé, (Villejuif Cedex, France):
Involvement of non MDs in genetic service in France;
Gwen Anderson (Lincoln Center, MA, USA):
Genetics and nursing research, education, and practice in the 21st century;
Ann Happ Boldt, (Indianapolis, IN, USA):
The Evolution of the Genetic Counseling Profession in the United States;
Karen Greendale, (Albany, NY, USA):
The National Society of Genetic Counselors: Past, Present, and Future;
Penny Guilbert (Nottingham, UK):
Defining the skills and knowledge base of genetic nursing practice;
Gioia Iacopini, (Rome, Italy):
Organization of genetic counseling in Italy: input of psychosocial disciplines;
Lauren Kerzin-Storrar, (Manchester, UK):
The Manchester MSc genetic counselling course: five year's experience;
Aad Tibben (Rotterdam, the Netherlands):
Genetic counseling: a matter of personality or skills?

Scientific Commitee:
Ségolène Aymé (Villejuif Cedex, France)
Gwen Anderson (Lincoln Center, MA, USA)
Ann Happ Boldt (Indianapolis, IN, USA)
Romeo Carrozzo (Milan, Italy)
Domenico A. Coviello (Genoa, Italy)
Franca Dagna-Bricarelli (Genoa, Italy)
Penny Guilbert (Nottingham, UK)
Gioia Iacopini (Rome, Italy)
Lauren Kerzin-Storrar (Manchester, UK)
Aad Tibben (Rotterdam, the Netherlands)

Organizing Committee:

Romeo Carrozzo (Milan, Italy)
Domenico A. Coviello (Genoa, Italy)
Franca Dagna-Bricarelli (Genoa, Italy)

Registration fee:
Lire 100.000

For information please contact:
Dr. Domenico A. Coviello, M.D., Ph.D.
Institute of Biology and Genetics,
University of Genoa,
Viale Benedetto XV,6; 16132-Genoa, Italy.
Tel. +39-10-353.8980; Fax +39-10-353.8978
e-mail: coviello@unige.it

Latebreaking Presentations:

Please send a one page summary (in accordance with the abstract submission instructions), together with a covering letter explaining its importance, before April 24, 1997 to Dr. Coviello

ORGANISATION OF GENETIC SERVICES IN ITALY:
INPUT OF PSYCHOSOCIAL DISCIPLINES.

JACOPINI,A.G.(1),FRONTALI,M.(2),
PEROBELLI,S.(3),SALMASO,D.(1)

1.Istituto di Psicologia, CNR - Roma

2.Istituto Med.Sperimentale, CNR -Roma

3.Servizio Psicologia Clinica,
Centro Fibrosi Cistica - Verona

Introduction

At present, the main medical application of DNA technologies is in the diagnosis of genetic disorder or in the detection of increased susceptibility to disease: effective treatment is rare. Nevertheless, genetic tests are already influencing all aspects of medical practice.

In our opinion this stresses the need for genetic and psychosocial counseling of the highest standards because of the medical, psychosocial, ethical and legal problems raised by:

- the difficulties in offering, providing and interpreting predictive genetic tests;
- the predictive nature of much genetic information;
- the psychosocial impact of knowledge of genetic risk for the individual;
- the implications for other family members;
- the difficult decisions that people at risk often have to make.

In Italy there are various genetic diagnostic and counseling centres even though they are not uniformly distributed throughout the Country. We have asked ourselves just how many of these centres are truly capable of facing the complex psychosocial problems posed by genetic counseling in general and, in particular, by the management of diagnostic and predictive tests.

Questionnaires soliciting information from directors of genetic services were mailed to all the listings in the Guide to the Diagnostic and Treatment Services for Genetic Disorders that is the most complete and uptodate directory available in Italy.
A covering letter, accompanying each questionnaire, outlined the purpose of the study as desiring to determine how the genetic centres were organised and which services were delivered (laboratory diagnostic procedures only, or genetic counseling or both).

The questionnaire was divided into 2 parts.
Part A of the questionnaire was to be filled in by the person in charge of the service.
Part B of the Questionnaire was to be filled in by the person who dials with the psychosocial aspects of g.c.

We mailed 193 questionnaires.

The results we are presenting here are not complete in that not all the recipients have answered so far and we have limited this presentation to the 82 centres whose responses have come within April 30. Other responses have come later and are still coming and we will include all of them in the final report.

However, though being not complete, the data we present here do trace a realistic picture of the genetic services in our Country as they came from 17 out of the 19 Italian Regions, hence they cover a fairly large proportion of the national territory.

QUESTIONNAIRE A - THE ORGANISATION OF G.C.

95% of the centres that have answered so far carry out diagnostic activity;

82% is the proportion of those carrying out both diagnostic and counseling activities;

Only 65% of the centres provides the counseling when the diagnostic results are given.

We do not know the characteristics of the centres that do not always provide counseling at the time of diagnosis: it is possible, for instance, that they hand over the diagnosis to the doctor and not to the patient.

However, it is a rather important issue and needs to be further investigated both because ethical guidelines tend to attribute the responsibility of the counseling to the centre that performs the test and also because it could indicate an underevaluation of the psychological impact of the diagnostic or predictive genetic information.

Who deals with the psychosocial aspects ?

33% of the centres did not answer;

54.5% of the respondents stated that this task is carried out by the doctors or by the biologists;

45.5% assigns the task of managing the psychosocial problems to social workers or to psychologists, either alone (34.5%) or together with the medical staff (11%).

Amongst the doctors who take care of the psychological aspects only 11% have taken ad hoc courses.

81% of the doctors that have responded deem that they would need some specific training as they feel not to be capable of living up to expectations.

The training of the medical staff is evaluated as insufficient to deal with such issues as:

- adaptation to/acceptance of the disease;

- social/employment-related difficulties experienced by the patient;

- anxiety and feeling of guilt that come with the diagnosis;

- difficulties in making reproductive choices;

- complex family and couple dynamics.

Doctors also indicate as very difficult:

- the communication of the diagnosis;

- the explanation of the concept of genetic risk.

11.2% of the centres that have responded can rely on psychosocial operators employed on a permanent or temporary basis in the service .

25.8% seek for creative solutions such as:

- using staff that is not specifically trained
  for working in a genetic centre, namely the psychologists of family counseling centres or medical psychiatrists who work in the establishment.
- recoursing to provisional, because often unpaid, collaborators, namely external psychologists or psychiatrists.
- allying with psychologists from other institutions on the basis of scientific collaboration agreements.

63% of the respondents, however, do not have any psychosocial operator available to them.

We tryed to find out whether there might be some specific features differentiating the 23 centres that have organised themselves in one way or another to be able to give some psychosocial assistance.

There don't appear to be any geographic differences.

Instead there do appear to be some differences where the types of diseases dealt with in these centres are taken into account.

The genetic centres endowed with some form of psychosocial service deal with individual diseases such as, for instance, c.f. or with uniform groups of genetic diseases, such as neuromuscular disorders.

Conversely, the genetic centres without any psychosocial assistance mostly provide services for all disorders.

In our opinion, one of the reasons accounting for more attention being paid to psychosocial problems by such centres as those for c. f. and for neuromuscular disorders, is the close co-operation between the centres and the lay Associations for these diseases, which also happen to be the most frequently found genetic disorders.

Of course, the lay Associations succeed in having a greater impact on the organisation of those genetic centres which specialize in only one disease, or in a uniform group of disorders.

In these cases lay Associations can be very influential in:

- linking research and clinical practice with the family and social settings of the patients;
- informing the families about the new findings in Genetics and Pharmacologies;
- contributing to educating society, abating prejudice and promoting solidarity with the patients by spreading information about the diseases;
- encouraging research also by raising funds.

(75% of the centres endowed with psycho-social staff entertain close collaboration with such associations; whereas

43% of the centres without psycho-social staff report only intermittent co-operation with lay associations).

QUESTIONNAIRE B - THE PSYCHOSOCIAL OPERATORS

None of the respondents worked with the genetic centres before 1979.

Most of them (57.7%) have begun to work in the area of genetic counseling only since 1990.

These operators are mostly called in on a routine basis (88%)

Less than half (38.4%) have taken specific training courses on g.c. and all but one feel that specific training is indispensable.

The aspects of professional training felt to be specific and necessary are:

knowledge of genetics

knowledge of risk evaluation

knowledge of the characteristics of genetic diseases

ability to analyse complex situations and relations, as those for which genetic diseases are responsible

ability to intervene in a crisis situation.

In about 85% of the cases the assessments made by the psychosocial operator are discussed with the team of consulting physicians, and however the respondents complain about:

- the lack of coordination between him/her self and the doctors
- directiveness of the team of physicians
- insufficient exploration of the psychosocial problems
- lack of boundaries in professional accountability

CONCLUSIONS

Genetic information has profound implications for individual and society. The genetic counseling context is one in which decisions must be made on how to best integrate this information with historical, personal and social understandings.

The key psychological concept is that of "meaning", that is the personal, subjective significance of the total information received by the individual or couple.

Relative unfamiliarity with the dynamics or processes of personal counseling tend, on one hand, to lead to an underestimate of the degree to which counseling principles influence practice and procedures and, on the other, to overestimate the kind of psychosocial interventions that are needed in genetic counseling.

Some respondents seem to imply that the usual problems encountered in genetic counseling require interventions and models more appropriate to psychopathology than to normal behavior, while others (11) seem to imply that counseling skills can be acquired on the job.

This belief is naive and unrealistic. Experiences in other areas of personal counseling suggest that the acquisition of even the rudiments of effective listening, responding and non directiveness is not as simple as it seems.

At present, as far as we know, genetic testing in this Country has been developing without harm because testing is undertaken in genetic and/or research institutes, in consultation with geneticists and trained psychologists.

In the next few years, however, the use of genetic testing is likely to expand rapidly outside research institutes, in the "real" world we have just described.

This means that a major role in the provision of genetic testing will be played by primary health care providers and other non-genetic specialists who have had only general notions about Genetics and no training whatever with regard to bioethics, or personal counseling, or doctor-patient interaction other than the traditional relationship acquired within the framework of therapeutic medicine.

There is plenty of evidence that the problems in providing genetic tests are different from those encountered in other areas of medical practice.

In actual fact, the predictive use of genetic tests places the doctor before a healthy patient and he/she should provide a setting conducive to an autonomous decision without taking on an authoritative attitude. This is the direct opposite of the conduct that most physicians have, who are used to *taking on therapeutic responsibility for their patient.*

The data clearly show that in this Country the Health, National and Regional, authorities have been very slow in planning the Genetic Counseling Services and in making available ad hoc personnel to meet the needs that were being prompted by research in the field of genetic tests.

We think that the data we have collected underline the need for:

- promoting educational programs in order to make medical students, health care providers and all personnel involved in referring patients more knowledgeable about genetics and risks, benefits and limitations of genetic tests so to ensure adequate incorporation of genetic testing into health care delivery system.

- establishing training course for non-medical genetic counselors who are experts in the management of psychosocial problems related to genetic counseling;

- promoting health education programmes aimed at enabling the general population to make their way through the jungle of incorrect information that abounds in this sector.

Indeed, in order for the users of genetic tests to be able to exercise autonomous decision-making it is necessary for them to upgrade and expand their basic medical, scientific and genetic knowledge.